spondyloperipheral dysplasia

Spondyloperipheral dysplasia is a disorder that impairs bone growth. This condition is characterized by flattened bones of the spine (platyspondyly) and unusually short fingers and toes (brachydactyly), with the exception of the first (big) toes. Other skeletal abnormalities associated with spondyloperipheral dysplasia include short stature, shortened long bones of the arms and legs, exaggerated curvature of the lower back (lordosis), and an inward- and upward-turning foot (clubfoot). Additionally, some affected individuals have nearsightedness (myopia), hearing loss, and intellectual disability.

Frequency

This condition is rare; only a few affected individuals have been reported worldwide.

Genetic Changes

Spondyloperipheral dysplasia is one of a spectrum of skeletal disorders caused by mutations in the *COL2A1* gene. This gene provides instructions for making a protein that forms type II collagen. This type of collagen is found mostly in the clear gel that fills the eyeball (the vitreous) and in cartilage. Cartilage is a tough, flexible tissue that makes up much of the skeleton during early development. Most cartilage is later converted to bone, except for the cartilage that continues to cover and protect the ends of bones and is present in the nose and external ears. Type II collagen is essential for the normal development of bones and other connective tissues that form the body's supportive framework.

Mutations in the *COL2A1* gene interfere with the assembly of type II collagen molecules, reducing the amount of this type of collagen in the body. Instead of forming collagen molecules, the abnormal COL2A1 protein builds up in cartilage cells (chondrocytes). These changes disrupt the normal development of bones and other connective tissues, leading to the signs and symptoms of spondyloperipheral dysplasia.

Inheritance Pattern

This condition is probably inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- SPD
- spondyloperipheral dysplasia with short ulna

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Spondyloperipheral dysplasia
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0796173/

Other Diagnosis and Management Resources

- MedlinePlus Encyclopedia: Nearsightedness
  https://medlineplus.gov/ency/article/001023.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Nearsightedness
  https://medlineplus.gov/ency/article/001023.htm
- Health Topic: Bone Diseases
  https://medlineplus.gov/bonediseases.html
- Health Topic: Connective Tissue Disorders
  https://medlineplus.gov/connectivetissuedisorders.html

Genetic and Rare Diseases Information Center

- Spondyloperipheral dysplasia
  https://rarediseases.info.nih.gov/diseases/4994/spondyloperipheral-dysplasia

Additional NIH Resources

- National Institute of Arthritis and Musculoskeletal and Skin Diseases
  https://www.niams.nih.gov/Health_Info/Connective_Tissue/

Educational Resources

- Disease InfoSearch: Spondyloperipheral dysplasia
  http://www.diseaseinfosearch.org/Spondyloperipheral+dysplasia/6843
- MalaCards: spondyloperipheral dysplasia
  http://www.malacards.org/card/spondyloperipheral_dysplasia
- Nemours Children's Health System: Skeletal Dysplasia
  https://www.nemours.org/service/medical/skeletal-dysplasia.html?tab=about
- Orphanet: Spondyloperipheral dysplasia-short ulna syndrome
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1856

Patient Support and Advocacy Resources

- Human Growth Foundation
  http://hgfound.org/
- International Skeletal Dysplasia Registry, UCLA
  http://ortho.ucla.edu/isdr
- Little People of America
  http://www.lpaonline.org/
- MAGIC Foundation
  https://www.magicfoundation.org/
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/connect.html

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28spondyloperipheral+dysplasia%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- SPONDYLOPERIPHERAL DYSPLASIA
  http://omim.org/entry/271700

Sources for This Summary

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/8723097
- Zankl A, Zabel B, Hilbert K, Wildhardt G, Cuenot S, Xavier B, Ha-Vinh R, Bonafé L, Spranger J, Superti-Furga A. Spondyloperipheral dysplasia is caused by truncating mutations in the C-propeptide of COL2A1. Am J Med Genet A. 2004 Aug 30;129A(2):144-8.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15316962

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/condition/spondyloperipheral-dysplasia

Reviewed: July 2008
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services